

Contrasting profiles of everyday executive function in Smith-Magenis syndrome and down syndrome

Wilde, Lucy; Oliver, Christopher

DOI:

[10.1007/s10803-017-3140-2](https://doi.org/10.1007/s10803-017-3140-2)

Document Version

Peer reviewed version

Citation for published version (Harvard):

Wilde, L & Oliver, C 2017, 'Contrasting profiles of everyday executive function in Smith-Magenis syndrome and down syndrome', *Journal of Autism and Developmental Disorders*. <https://doi.org/10.1007/s10803-017-3140-2>

[Link to publication on Research at Birmingham portal](#)

Publisher Rights Statement:

Checked for eligibility: 23/05/2017

The final publication is available at Springer via <http://dx.doi.org/10.1007/s10803-017-3140-2>
<https://link.springer.com/article/10.1007%2Fs10803-017-3140-2>

General rights

Unless a licence is specified above, all rights (including copyright and moral rights) in this document are retained by the authors and/or the copyright holders. The express permission of the copyright holder must be obtained for any use of this material other than for purposes permitted by law.

- Users may freely distribute the URL that is used to identify this publication.
- Users may download and/or print one copy of the publication from the University of Birmingham research portal for the purpose of private study or non-commercial research.
- User may use extracts from the document in line with the concept of 'fair dealing' under the Copyright, Designs and Patents Act 1988 (?)
- Users may not further distribute the material nor use it for the purposes of commercial gain.

Where a licence is displayed above, please note the terms and conditions of the licence govern your use of this document.

When citing, please reference the published version.

Take down policy

While the University of Birmingham exercises care and attention in making items available there are rare occasions when an item has been uploaded in error or has been deemed to be commercially or otherwise sensitive.

If you believe that this is the case for this document, please contact UBIRA@lists.bham.ac.uk providing details and we will remove access to the work immediately and investigate.

Abstract

Everyday executive function (EF) was examined in Smith-Magenis syndrome (SMS), associated with high risk of behaviour disorder, and Down syndrome (DS), associated with relatively low risk of behaviour disorder. Caregivers of 13 children with SMS and 17 with DS rated everyday EF using the Behavioral Rating Inventory of Executive Functioning-Preschool (BRIEF-P). Greater everyday EF deficits relative to adaptive ability were evident in SMS than in DS. The SMS profile of everyday EF abilities was relatively uniform; in DS emotional control strengths and working memory weaknesses were evident. Findings implicate broad everyday EF difficulties in SMS compared to DS, corresponding with increased rates of behaviour disorder in SMS. Findings further suggest that everyday EF profiles may, in part, be syndrome related.

EVERYDAY EF IN SMITH-MAGENIS AND DOWN SYNDROMES

Executive functions (EFs) are abilities relating to higher order self-regulatory processes, including planning, set-shifting, inhibition, working memory and cognitive flexibility (Griffith et al. 1999). Some behaviours considered phenotypic of genetic neurodevelopmental disorders may be explained by specific executive function deficits. For example in Down syndrome (DS) and Williams syndrome, deficits in response inhibition have been hypothesised to underpin high levels of social approach behaviour (Porter et al. 2007). When genetic neurodevelopmental disorders evidence contrasting behavioural phenotypes, divergent underlying profiles of EF abilities might therefore be anticipated. A diverse literature links EF deficits to adverse behavioural outcomes. For example, symptoms of psychiatric disorders such as ADHD and obsessive compulsive disorder are associated with deficits in a range of EF abilities (Snyder et al. 2015; Willcutt et al. 2005). Poorer EF abilities are also evidenced by individuals with antisocial behaviour (Morgan & Lilienfeld, 2000) and damage to the frontal lobes, the area of the brain which subserves EF, also results in behaviour problems including impulsivity and irritability (McAllister, 2008).

Two genetic neurodevelopmental disorders which evidence contrasting profiles of behaviour disorder, and therefore potentially divergent EF profiles, are Smith-Magenis syndrome (SMS), a rare syndrome caused by either a deletion of chromosome 17p11.2 or mutation of the retinoic acid-induced 1 (RAI1) gene (Greenberg et al. 1991; Slager et al. 2003), and Down syndrome (trisomy 21). The behavioural phenotype of SMS encompasses a diverse range of behaviour problems. Self-injury is described as near universal, reported in 93% of individuals, and aggression is reported in 74% of individuals (Arron et al. 2011). This is much higher than in the wider intellectual disability (ID) population (Emerson et al. 2001) or other genetic syndromes associated with ID (Arron et al. 2011). High levels of impulsivity and a wide range of repetitive behaviours are described (Moss et al. 2009; Oliver et al. 2011).

In contrast, descriptions of the behavioural phenotype of DS implicate relatively low rates of behaviour disorder. Self-injury and aggression are reported in just 7.22% and 8.61% of individuals, significantly less than age-matched peers with nonspecific ID and lower rates of a wide range of other behaviour problems, and fewer psychiatric disorders, are evident in DS (Collacott et al. 1998). Fewer behaviour problems are also evidenced relative to other genetic neurodevelopmental disorders (Walz & Benson, 2002). However, while DS is at lower risk for psychopathology relative to others with ID, there is still increased risk of behaviour problems compared to age-matched TD peers (Cuskelly & Dadds, 1992; Stores et al. 1998).

While SMS and DS differ notably in terms of problem behaviour there are similarities between the syndromes. Both are associated with, typically moderate, ID (Carr, 2012; Melyn & White, 1973; Osório et al. 2012; Udwin et al. 2001) and evidence expressive language weaknesses relative to receptive language (Gropman et al. 2006; Martin et al. 2009). Adaptive functioning profiles in both syndromes include socialisation strengths relative to communication (Fidler et al. 2006; Martin et al. 2006) and unusually strong social motivation is reported in both SMS and DS (Dykens & Smith, 1998; Kasari et al. 1990). Yet despite similarities in other relevant domains such as ability, SMS and DS clearly differ in the extent to which they are characterised by behaviour disorder. The current study therefore focussed on examining EF in these two syndromes because a diverse body of evidence points to this being a likely source of variability in behaviour disorder.

Despite possible associations with behavioural problems, while the SMS phenotype is increasingly well described, to date EF has not been specifically examined although there is suggestion of working memory weaknesses based on performance on IQ assessments (Osório et al. 2012). Conversely, a considerable body of evidence describes EF in DS. Performance based measures indicate deficits relative to mental age matched typically developing (TD) peers and peers with ID, in both adult and child/adolescent samples across a broad range of

EF domains, including particularly robust working memory difficulties (Baddeley & Jarrold, 2007; Lanfranchi et al. 2010; Rowe et al. 2006). Informant report of EF abilities in children/adolescents with DS also indicates deficits compared to TD individuals, across a range of EF abilities (Daunhauer et al. 2014; Lee et al. 2015; Lee et al. 2011), but interestingly relative strengths in emotional control are consistently evident in these studies, including when compared to others with a genetic syndrome (sex chromosome trisomy). The EF captured in informant report has been referred to as ‘everyday’ EF, reflecting the skills needed for individuals to get along in everyday life (Daunhauer et al. 2014). Toplak et al. (2013) suggest that performance and informant report measures assess different domains which contribute independently to clinical problems. Use of informant report of EF therefore potentially provides valuable insight into difficulties experienced by individuals with SMS and DS.

In summary, the majority of individuals with SMS evidence adverse behavioural outcomes; EF deficits may contribute to increased risk for such outcomes, but the profile of EF in this syndrome has not yet been described. Identification of everyday EF deficits in SMS may further understanding of factors associated with behavioural difficulties evident in the syndrome and potentially facilitate targeted intervention. Furthermore, the extent to which profiles of everyday EF described in informant report in DS are syndrome specific is unclear. Everyday EF abilities in DS have been compared to another genetic syndrome only by Lee et al. (2015) and in this study the contrast group did not have ID. Contrast between SMS and DS, both syndromes associated with ID, will therefore contribute to understanding of whether the profile of strengths and weaknesses in everyday EF may be to some extent syndrome related, or whether it is more likely only associated with ID.

The aim of this study was therefore to examine syndrome related profiles of caregiver reported everyday EF in SMS in contrast to DS. Given higher rates of behaviour disorder in

SMS than DS, and associations between EF deficits and behaviour problems, greater everyday EF deficits across a range of EF domains were anticipated in SMS than in DS. However, given evidence of working memory deficits in both SMS and DS, no between syndromes differences were anticipated in this domain. Within syndrome weaknesses in working memory were hypothesised in both groups, and in DS within syndrome strengths in emotional control were anticipated.

Methods

Participants and recruitment

Participants were a subset of children participating in a larger study of behaviour in SMS and DS (see [withheld for blind review] for details of full sample). Participants were recruited from the main UK based family support groups (Smith-Magenis Foundation UK and the Down's Syndrome Association) and an existing participant database held by [withheld for blind review]. Inclusion criteria were confirmed diagnosis of the genetic syndrome from an appropriate professional (e.g. clinical geneticist, paediatrician), age between 2 - 16 years and age equivalent scores on Vineland Adaptive Behavior Scales II (VABS II; Sparrow et al. 2005) between 2-5 years, the age range of the everyday EF measure used in the current study, the BRIEF-P (Gioia et al. 2003). The VABS II represents the most complete, and therefore most representative, measure of ability for the sample; data on cognitive ability was available for only around half of the sample due to either non-compliance or scoring at floor on the measure.

Seventeen children with DS and 13 with SMS met inclusion criteria. Demographic details are shown in Table 1. All children with DS had trisomy 21, 12 children with SMS had a chromosome 17p11.2 deletion and one had a gene *RAI1* mutation. No significant differences

EVERYDAY EF IN SMITH-MAGENIS AND DOWN SYNDROMES

were found between groups in gender, chronological age, mean age equivalents or mean standard score on the VABS.

+++++[Insert table 1 about here]+++++

Measures

Demographic Questionnaire. This provided information on date of birth, gender, mobility, verbal ability and diagnostic status.

Behavior Rating Inventory of Executive Function-Preschool (BRIEF-P, Gioia et al. 2003). This 63 item informant report measure of EF in ecologically valid contexts is designed for use with children aged from 2 years to 5 years 11 months. Caregivers rate the frequency of everyday behaviours purported to be associated with executive functioning deficits over the past six months across five subscales; Inhibit (inhibiting behavioural responses), Shift (shifting attention from one task to another), Emotional Control (regulation of emotional responses to everyday situations), Working Memory (retention and manipulation of information in everyday life) and Plan/Organize (planning and preparing for the future). Frequency of behaviour is rated as 1 (never), 2 (sometimes) or 3 (often). Three broader indices and a single composite score can also be derived. This preliminary examination focuses on the clinical scales as these offer the most specific information about EF domains (the indices and composite are derived directly from the clinical scales). Internal consistency, reliability and test-retest reliability are good and convergent and discriminant validity are robust (Sherman & Brooks, 2010).

The BRIEF-P was used in the current study in preference to the chronological-age appropriate BRIEF (Gioia et al 2000) because it is likely to assess skills which are developmentally appropriate for children and adolescents with ID. Were participants assessed

using the chronological-age appropriate BRIEF, the majority would score at or near ceiling on impairments of everyday EF and analyses may then not be sensitive to different profiles of everyday EF.

Vineland Adaptive Behavior Scales – Interview edition, Survey form (VABS II -SF, Sparrow et al., 2005). This semi structured interview with carers consists of 261 items assessing adaptive abilities; the practical, conceptual and social skills learnt and performed in everyday life (Schalock et al. 2010). Items form four domains (sub domains noted in brackets); Communication (Receptive, Expressive and Written), Daily-living Skills (Personal, Domestic, Community), Socialisation (Interpersonal Relationships, Play and Leisure Time, Coping Skills) and Motor Skills (Fine and Gross). Age-equivalents can be calculated for each sub-domain and standard scores can be calculated for each domain. The scales have high test-retest and inter-rater reliability, and internal consistency for each domain is good (Sparrow et al. 1984).

Using the VABS II as the measure of ability in the current study enables assessment of everyday EF in the context of individuals' level of adaptive ability. Adaptive ability deficits are a diagnostic feature of ID and there is increasing recognition of the importance of considering adaptive abilities when evaluating functioning in individuals with ID (Tassé et al. 2012). Adaptive abilities may also be particularly pertinent to functioning in everyday contexts, which are the focus of the current study.

Procedure

Ethical review and approval was obtained from the ethics committee of the [withheld for blind review]. Caregivers received questionnaire packs (cover letter, information sheet, questionnaire pack, consent form and a prepaid envelope) on the day of an assessment visit

and either returned them on the assessment day or by post. VABS data were gathered either shortly before or after the research visit, over the telephone.

Data analysis

Raw BRIEF-P scale scores were converted to normative T scores (age and gender referenced), calculated the mean age equivalent of the VABS subdomains. This approach (deriving T scores using estimates of developmental, rather than chronological, age) accounts for developmental delay, enabling identification of everyday EF deficits beyond those expected from a child's overall developmental level (Daunhauer et al. 2014). In the current study age equivalents from the VABS II, a measure of adaptive ability, were used to derive T scores; this enables consideration of whether impaired scores on the BRIEF-P indicate everyday EF difficulties in excess of overall adaptive delays.

These T scores were compared between syndromes for each clinical scale, within syndromes (comparing pairs of clinical scales) and also to the normative mean score for TD children of 50 (separately for each syndrome). Shapiro-Wilk tests indicated data for T scores were normally distributed ($p > .05$), except for the Emotional Control scale, which was significantly non-normally distributed for both syndrome groups ($p < .05$). Therefore, parametric analyses were employed and for analyses involving non-normally distributed data supplementary non-parametric alternatives were also used where possible. Bonferroni corrections were employed for multiple comparisons.

Results

Figure 1 shows the profile of mean T scores for children with SMS and DS, together with the mean normative T score of 50. A mixed measures ANOVA examined the difference between syndrome groups on the pattern of T scores across the clinical scales (Greenhouse-Geisser

values reported due to violations of sphericity). Main effects of clinical scale ($F(2.63, 70.89) = 26.16, p < .001$) and of syndrome group ($F(1, 27) = 15.03, p = .001$) were found. A significant interaction between clinical scale and group ($F(2.63, 70.89) = 3.93, p = .015$) indicated that the pattern of T scores on the clinical scales differed significantly between SMS and DS.

+++++[Insert figure 1 about here]+++++

Table 2 presents the descriptive statistics for the BRIEF-P T scores for each syndrome. Between groups t tests indicated that children with SMS showed more impairments than children with DS on the Inhibit ($t(28) = 2.87, p = .008, d = 1.04$), Shift ($t(28) = 2.89, p = .007, d = 1.07$) and Emotional Control ($t(28) = 5.34, p < .001, d = 1.96$)¹ scales, but not the Working Memory ($t(27) = 1.53, p = .138, d = 0.56$) or Plan/Organize scales ($t(17.647) = 1.88, p = .077, d = 0.72$).

+++++[Insert table 2 about here]+++++

In terms of within syndromes patterns of strengths and weaknesses in everyday EF abilities, within subjects t tests (shown in in table 3) indicated that in SMS there were impairments only in in Working Memory relative to both Shift and Emotional Control² and in Inhibit relative to Shift. In DS there were impairments in working memory relative to Inhibit, Shift, Emotional Control and Plan/Organize, but fewer impairments in Emotional Control³ compared to Inhibit, Plan/Organize and Working Memory. The trend towards greater impairments in Plan/Organize compared to Shift should also be noted as it was associated with a large effect size.

¹ This difference was also significant in Mann-Whitney analysis, $U = 17.5, p < .001$

² This difference was also significant in a Wilcoxon analysis $Z = -2.54, p = .011$

³ These differences were also significant in a Wilcoxon analysis; Inhibit $Z = -3.43, p = .001$, Plan/Organize $Z = -3.32, p = .001$ and Working Memory $Z = -3.62, p < .001$ but not Shift $Z = -2.38, p = .017$.

+++++[Insert table 3 about here]+++++

When compared to the normative mean T score of 50, children with SMS had higher mean T scores (denoting greater difficulties) on all clinical scales of the BRIEF-P (Inhibit $t(12) = 7.85, p < .001, d = 2.43$; Shift $t(12) = 5.91, p < .001, d = 1.66$; Emotional Control $t(12) = 7.11, p < .001, d = 2.07$; Working Memory $t(12) = 8.57, p < .001, d = 2.73$; Plan/Organize $t(12) = 5.64, p < .001, d = 1.82$), whereas children with DS had higher mean T scores for Inhibit ($t(16) = 7.29, p < .001, d = 1.70$), Working Memory ($t(16) = 10.11, p < .001, d = 1.56$) and Plan/Organize ($t(16) = 7.26, p < .001, d = 2.46$), but not Shift ($t(16) = 1.92, p = .073, d = 0.49$) or Emotional Control ($t(16) = .40, p = .693, d = 0.10$).

Examination of effect sizes for all t -test analyses indicates that all large effects (.8; Cohen, 1988) were associated with a significant difference, except the Shift-Plan/organize within syndrome comparison for DS, suggesting that overall analyses did not fail to identify large effects despite small sample sizes.

Discussion

This study was the first to contrast syndrome related profiles of everyday EF difficulties in genetic neurodevelopmental disorders associated with ID. It was anticipated that as SMS and DS have contrasting behavioural phenotypes, primarily in terms of behaviour disorder, profiles of everyday EF would also be divergent, with greater difficulties evident in SMS than DS. Findings supported this, with a main effect indicating greater everyday EF deficits when adaptive functioning levels were taken into account, in SMS than in DS. An interaction indicated that the extent of the differences between syndromes varied according to the everyday EF domain being assessed. Taking into account adaptive functioning there were broad everyday EF difficulties in SMS, resulting in a relatively flat profile in SMS, whereas

in DS clear working memory deficits and emotional control strengths were evident. The broad EF deficits in SMS were also evident in comparison to TD norms, whereas in DS no differences were found for two of the five everyday EF domains assessed.

The behaviour problems in SMS, including impulsivity, self-injury and aggression, temper outbursts and repetitive/stereotyped behaviour (Dykens et al. 1997; Moss et al. 2009; Sloneem et al. 2011), implicate problems with inhibiting behavioural and emotional responses and in shifting attention. In the current study these were also abilities for which deficits, relative to expectations based on adaptive functioning abilities, were identified for children with SMS compared to children with DS. The correspondence suggests that difficulties with these everyday EF abilities could potentially underpin some of the behaviour problems that differentiate SMS and DS. Failure of children with SMS in the current study to differ on the working memory subscale from those with DS (for whom there is very clear evidence of working memory difficulties) offers further support for the suggestion of specific working memory difficulties in SMS, as proposed by Osório et al. (2012). The deficits evident in SMS across all everyday EF domains when compared to TD norms, further suggests that relative to children without ID of similar adaptive ability there are significant problems with the broad set of abilities involved in control and regulation of behaviour.

The within syndrome profile of abilities in SMS was not characterised by clear strengths and weaknesses; working memory was the only everyday EF domain to differ from more than one other BRIEF-P domain. The suggestion of relative working memory deficits (in comparison to other abilities in SMS), as well as an absolute deficit (compared to peers of a similar developmental age without ID) is consistent with past research. Using the Wechsler intelligence scales, Osório et al. (2012) found working memory weakness not only relative to TD controls, but mean working memory scores of individuals with SMS were lower than their mean verbal comprehension index scores. It is perhaps surprising that specific

difficulties with everyday inhibition of behaviour were not evident in SMS. One of the most unusual features of SMS is extremely high prevalence of self-injury, and models of self-injury implicate inhibition deficits in both the development and maintenance of self-injurious behaviour (Oliver & Richards, 2015). Examination of the association between inhibition and self-injury warrants further investigation in SMS

The within syndrome profile of everyday EF found for DS in the current study is highly consistent with past research (Lee et al. 2011), suggesting that everyday EF in DS may be robustly characterised by weaknesses in working memory and strengths in emotional control. Concordance between informant report of everyday working memory and performance measures further suggests that working memory deficits in DS manifest across diverse contexts. Within syndrome strengths in emotional control with respect to adaptive functioning ability, in conjunction with similarities to emotional control scores of TD children and the finding in the current study of better emotional control than another group of children with a genetic syndrome associated with ID when adaptive functioning ability is taken into account, suggests that this ability may be ‘spared’ in DS when developmental level (either intellectual or adaptive functioning development) is taken into account. In the current study shifting standard scores in children with DS were also not found to differ from those of TD children, replicating the findings of Daunhauer et al. (2014), thus behavioural flexibility may also be preserved in DS when developmental age is taken into account. However, other studies have not found such strengths in either within or between group comparisons (Lee et al. 2015; Lee et al. 2011), therefore this is a less robust area of strength in DS.

Drawing between and within syndrome findings together, while both SMS and DS evidence everyday working memory difficulties, in SMS there are additional difficulties across multiple everyday EF domains, whereas in DS relative strengths may exist, specifically in emotional control. Therefore, in terms of the question of whether contrasting underlying

profiles of EF abilities might account for behavioural phenotypes which diverge in terms of problematic behaviours, being able to regulate emotional responses may be protective against development of a range of behaviour disorders. Conversely, where emotional regulation difficulties exist, particularly in conjunction with additional self-regulation deficits, increased risk of behaviour disorder may be conferred.

Future research would therefore benefit from more focussed examination of the association between specific everyday EF deficits, including emotional control, and specific phenotypic behaviour problems in genetic neurodevelopmental disorders. Self-injury in SMS, which is of obvious clinical significance, is a clear candidate for such investigation. Similarly it is evident that working memory is a prominent difficulty in DS, but it is not clear if there are any consequences of this in terms of behaviour disorder. Future studies of everyday EF would also benefit from assessing cognitive, as well as adaptive, functioning to enable evaluation of deficits in everyday EF relative to both adaptive and intellectual ability, providing a more complete consideration of the impact of ID. Larger sample sizes would also be beneficial, particularly for the SMS group (where novel preliminary findings suggest difficulties that should be examined in further detail) to increase power and improve ability to match across syndrome groups on measures of ability and age (the mean age of the SMS group was greater than the DS group, thus the SMS group had more life experience than the DS group).

In summary, this first comparison of everyday EF in two genetic neurodevelopmental disorders associated with ID has identified divergent profiles between syndromes. Findings provide early indications of broad everyday EF deficits in SMS relative to adaptive ability and further elucidate profiles of everyday EF in DS, characterised by working memory difficulties and emotional control strengths relative to adaptive ability. In SMS interventions supporting a range of EF domains during everyday activities may therefore be beneficial,

perhaps focussing initially on working memory e.g. using visual timetables. In DS, working memory support is also clearly indicated. A wider implication of the study is that particular everyday EF profiles may to an extent be syndrome related, as opposed to solely being a characteristic of ID. However, because relative to TD peers of the same chronological age a broad deficit in all everyday EF domains would likely have been found, everyday EF profiles are also likely to be affected by presence of ID.

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all individual participants included in the study.

References

- Arron, K., Oliver, C., Moss, J., Berg, K., & Burbidge, C. (2011). The prevalence and phenomenology of self-injurious and aggressive behaviour in genetic syndromes. *Journal of Intellectual Disability Research*, 55(2), 109-120.
- Baddeley, A., & Jarrold, C. (2007). Working memory and Down syndrome. *Journal of Intellectual Disability Research*, 51(12), 925-931.
- Carr, J. (2012). Six weeks to 45 years: a longitudinal study of a population with Down syndrome. *Journal of Applied Research in Intellectual Disabilities*, 25(5), 414-422.
- Cohen, J. (1988), *Statistical Power Analysis for the Behavioral Sciences*, 2nd Edition. Hillsdale: Lawrence Erlbaum
- Collacott, R., Cooper, S., Branford, D., & McGrother, C. (1998). Behaviour phenotype for Down's syndrome. *The British Journal of Psychiatry*, 172(1), 85-89.

EVERYDAY EF IN SMITH-MAGENIS AND DOWN SYNDROMES

- Cuskelly, M., & Dadds, M. (1992). Behavioural Problems in Children with Down's Syndrome and their Siblings. *Journal of Child Psychology and Psychiatry*, 33(4), 749-761.
- Daunhauer, L. A., Fidler, D. J., Hahn, L., Will, E., Lee, N. R., & Hepburn, S. (2014). Profiles of everyday executive functioning in young children with Down syndrome. *American Journal on Intellectual and Developmental Disabilities*, 119(4), 303-318.
- Dykens, E. M., Finucane, B. M., & Gayley, C. (1997). Brief report: Cognitive and behavioral profiles in persons with Smith-Magenis syndrome. *Journal of Autism and Developmental Disorders*, 27(2), 203-211.
- Dykens, E. M., & Smith, A. C. M. (1998). Distinctiveness and correlates of maladaptive behaviour in children and adolescents with Smith-Magenis syndrome. *Journal of Intellectual Disability Research*, 42, 481-489.
- Emerson, E., Kiernan, C., Alborz, A., Reeves, D., Mason, H., Swarbrick, R., . . . Hatton, C. (2001). The prevalence of challenging behaviors: A total population study. *Research in Developmental Disabilities*, 22(1), 77-93.
- Fidler, D., Hepburn, S., & Rogers, S. (2006). Early learning and adaptive behaviour in toddlers with Down syndrome: evidence for an emerging behavioural phenotype? *Down Syndrome Research and Practice*, 9(3), 37-44.
- Gioia, G. A., Espy, K. A., & Isquith, P. K. (2003). *BRIEF-P: Behavior Rating Inventory of Executive Function-Preschool Version: Professional Manual*: Psychological Assessment Resources.
- Greenberg, F., Guzzetta, V., Deocaluna, R. M., Magenis, R. E., Smith, A. C. M., Richter, S. F., . . . Lupski, J. R. (1991). Molecular Analysis of the Smith-Magenis Syndrome - a Possible Contiguous-Gene Syndrome Associated with Del(17)(P11.2). *American Journal of Human Genetics*, 49(6), 1207-1218.

EVERYDAY EF IN SMITH-MAGENIS AND DOWN SYNDROMES

- Griffith, E. M., Pennington, B. F., Wehner, E. A., & Rogers, S. J. (1999). Executive functions in young children with autism. *Child Development*, 817-832.
- Gropman, A. L., Duncan, W. C., & Smith, A. C. M. (2006). Neurologic and developmental features of the Smith-Magenis syndrome (del 17p11.2). *Pediatric Neurology*, 34(5), 337-350.
- Kasari, C., Mundy, P., Yirmiya, N., & Sigman, M. (1990). Affect and attention in children with Down syndrome. *American Journal on Mental Retardation*, 95(1), 55-67.
- Lanfranchi, S., Jerman, O., Dal Pont, E., Alberti, A., & Vianello, R. (2010). Executive function in adolescents with Down Syndrome. *Journal of Intellectual Disability Research*, 54(4), 308-319.
- Lee, N. R., Anand, P., Will, E., Adeyemi, E. I., Clasen, L. S., Blumenthal, J. D., . . . Edgin, J. O. (2015). Everyday executive functions in Down syndrome from early childhood to young adulthood: evidence for both unique and shared characteristics compared to youth with sex chromosome trisomy (XXX and XXY). *Frontiers in Behavioral Neuroscience*, 9, 264.
- Lee, N. R., Fidler, D. J., Blakeley-Smith, A., Daunhauer, L., Robinson, C., & Hepburn, S. L. (2011). Caregiver Report of Executive Functioning in a Population-Based Sample of Young Children With Down Syndrome. *American Journal on Intellectual and Developmental Disabilities*, 116(4), 290-304.
- Martin, G. E., Klusek, J., Estigarribia, B., & Roberts, J. E. (2009). Language characteristics of individuals with Down syndrome. *Topics in Language Disorders*, 29(2), 112.
- Martin, S. C., Wolters, P. L., & Smith, A. C. M. (2006). Adaptive and maladaptive behavior in children with Smith-Magenis syndrome. *Journal of Autism and Developmental Disorders*, 36(4), 541-552.
- McAllister, T. W. (2008). Neurobehavioral sequelae of traumatic brain injury: evaluation and management. *World Psychiatry*, 7(1), 3-10.

- Melyn, M. A., & White, D. T. (1973). Mental and developmental milestones of noninstitutionalized Down's syndrome children. *Pediatrics*, 52(4), 542-545.
- Morgan, A. B., & Lilienfeld, S. O. (2000). A meta-analytic review of the relation between antisocial behavior and neuropsychological measures of executive function. *Clinical Psychology Review*, 20(1), 113-136.
- Moss, J., Oliver, C., Arron, K., Burbidge, C., & Berg, K. (2009). The Prevalence and Phenomenology of Repetitive Behavior in Genetic Syndromes. *Journal of Autism and Developmental Disorders* 39(4), 572-588.
- Oliver, C., Berg, K., Moss, J., Arron, K., & Burbidge, C. (2011). Delineation of behavioral phenotypes in genetic syndromes: characteristics of autism spectrum disorder, affect and hyperactivity. *Journal of Autism and Developmental Disorders*, 41(8), 1019-1032.
- Oliver, C., & Richards, C. (2015). Practitioner Review: Self-injurious behaviour in children with developmental delay. *Journal of Child Psychology and Psychiatry*, 56(10), 1042-1054.
- Osório, A., Cruz, R., Sampaio, A., Garayzábal, E., Carracedo, Á., & Fernández-Prieto, M. (2012). Cognitive functioning in children and adults with Smith-Magenis syndrome. *European Journal of Medical Genetics*, 55(6), 394-399.
- Porter, M. A., Coltheart, M., & Langdon, R. (2007). The neuropsychological basis of hypersociability in Williams and Down syndrome. *Neuropsychologia*, 45(12), 2839-2849.
- Rowe, J., Lavender, A., & Turk, V. (2006). Cognitive executive function in Down's syndrome. *British Journal of Clinical Psychology*, 45(1), 5-17.
- Schalock, R. L., Borthwick-Duffy, S. A., Bradley, V. J., Buntinx, W. H., Coulter, D. L., Craig, E. M., & Yeager, M. H. (2010). Intellectual Disability: Definition, Classification, and Systems of Supports. Washington. DC: *American Association on Intellectual and Developmental Disabilities*, 259.

- Slager, R. E., Lynn, T., Newton, T. L., Vlangos, C. N., Finucane, B., & Elsea, S. H. (2003). Mutations in RAI1 associated with Smith-Magenis syndrome. *Nature Genetics*, 33(4), 466-468.
- Sloneem, J., Oliver, C., Udwin, O., & Woodcock, K. A. (2011). Prevalence, phenomenology, aetiology and predictors of challenging behaviour in Smith-Magenis syndrome. *J Intellect Disabil Res*, 55(2), 138-151.
- Snyder, H. R., Kaiser, R. H., Warren, S. L., & Heller, W. (2015). Obsessive-compulsive disorder is associated with broad impairments in executive function: a meta-analysis. *Clinical Psychological Science*, 3(2), 301-330.
- Sparrow, S., Cicchetti, D., & Balla, D. (2005). Vineland adaptive behavior scales: (Vineland II), survey interview form/caregiver rating form. *Livonia, MN: Pearson Assessments*.
- Stores, R., Stores, G., Fellows, B., & Buckley, S. (1998). Daytime behaviour problems and maternal stress in children with Down's syndrome, their siblings, and non-intellectually disabled and other intellectually disabled peers. *Journal of Intellectual Disability Research*, 42(3), 228-237.
- Tassé, M. J., Schalock, R. L., Balboni, G., Bersani Jr, H., Borthwick-Duffy, S. A., Spreat, S., . . . Zhang, D. (2012). The construct of adaptive behavior: Its conceptualization, measurement, and use in the field of intellectual disability. *American Journal on Intellectual and Developmental Disabilities*, 117(4), 291-303.
- Toplak, M. E., West, R. F., & Stanovich, K. E. (2013). Do performance-based measures and ratings of executive function assess the same construct? *Journal of Child Psychology and Psychiatry*, 54(2), 131-143.
- Udwin, O., Webber, C., & Horn, I. (2001). Abilities and attainment in Smith-Magenis syndrome. *Developmental Medicine and Child Neurology*, 43(12), 823-828.

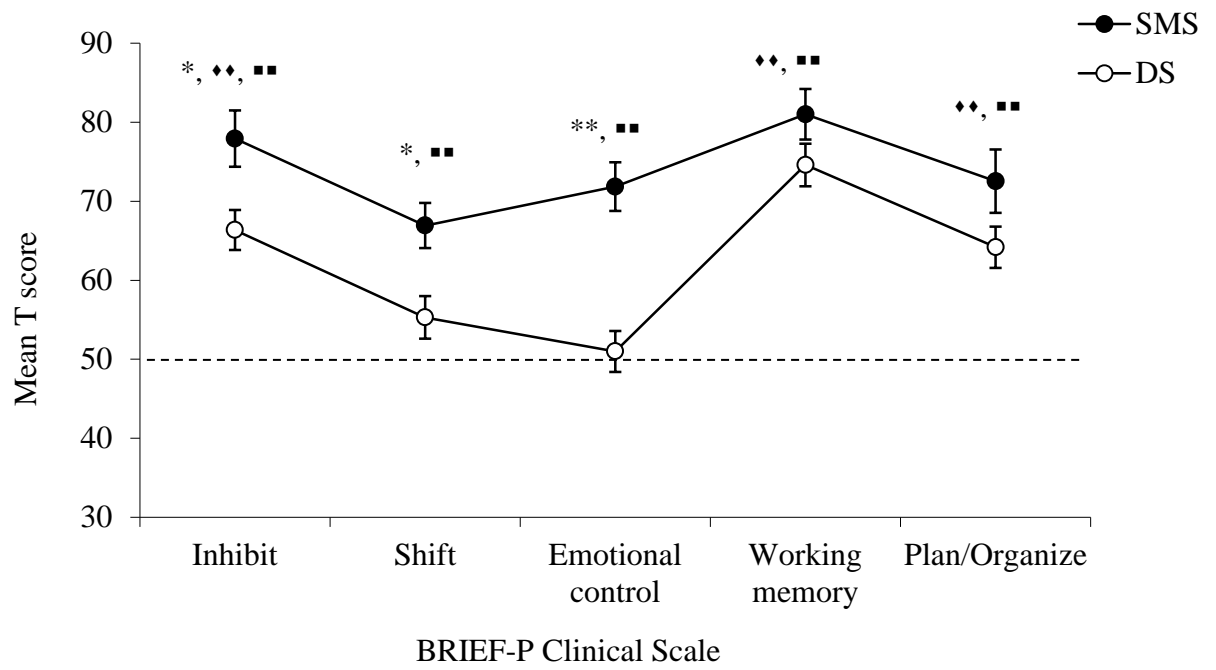
Walz, N. C., & Benson, B. A. (2002). Behavioral phenotypes in children with Down syndrome, Prader-Willi syndrome, or Angelman syndrome. *Journal of Developmental and Physical Disabilities, 14*(4), 307-321.

Willcutt, E. G., Doyle, A. E., Nigg, J. T., Faraone, S. V., & Pennington, B. F. (2005). Validity of the executive function theory of attention-deficit/hyperactivity disorder: a meta-analytic review. *Biological Psychiatry, 57*(11), 1336-1346.

Figure Captions

Figure 1. Profiles of caregiver reported everyday EF in Smith-Magenis syndrome (SMS) and Down syndrome (DS) on the BRIEF-P clinical scales. Dashed line represents mean normative T score (for DS and SMS comparisons * $p < .05$, ** $p < .005$, for DS and the mean normative T score comparisons ♦ $< .05$, ♦♦ $< .005$, for SMS and the mean normative T score comparisons, ■ $< .05$, ■■ $< .005$).

Figure 1 top



EVERYDAY EF IN SMITH-MAGENIS AND DOWN SYNDROMES

Table 1. Demographic characteristics of the participants. Mean age (standard deviation), number of participants who were males (percentage) and mean age equivalent⁴ and standard score across Vineland Adaptive Behavior Scales (VABS) subdomains/domains (standard deviation).

		Smith-Magenis syndrome	Down syndrome
N		13	17
Age (Months)	Mean	107.15	93.76
	(SD)	(46.74)	(32.26)
	Range	38-135	36-189
Gender	Number of males	7	9
	(%)	(53.8)	(52.9)
Mean adaptive behaviour age equivalent (Months)	Mean	42.81	42.00
	(SD)	(13.30)	(12.08)
	Range	29.89-67.22	25.56-70.44
Mean adaptive behaviour standard score	Mean	65.87	69.44
	(SD)	(9.81)	(7.53)
	Range	55.25-87.25	50.00-79.25

⁴ This is the mean of the subdomains described in the Measures subsection of this Method section.

EVERYDAY EF IN SMITH-MAGENIS AND DOWN SYNDROMES

Table 2. Mean, standard deviation and range of BRIEF-P scale standard scores for participants with Smith-Magenis syndrome and Down syndrome

BRIEF-P scale		Smith-Magenis	Down
		syndrome	syndrome
Inhibit	Mean	77.92	66.35
	(SD)	(12.82)	(9.25)
	Range	53-98	48-79
Shift	Mean	66.92	55.29
	(SD)	(10.33)	(11.36)
	Range	43-80	40-81
Emotional control	Mean	71.85	51.00
	(SD)	(11.07)	(10.24)
	Range	50-84	40-71
Working memory	Mean	81.00	74.59
	(SD)	(12.53)	(10.03)
	Range	50-97	56-89
Plan/Organize	Mean	72.54	64.18
	(SD)	(14.42)	(8.05)
	Range	51-94	49-81

EVERYDAY EF IN SMITH-MAGENIS AND DOWN SYNDROMES

Table 3. Within subjects comparisons of T scores on the BRIEF-P clinical scales for Smith-Magenis syndrome and Down syndrome (significant differences highlighted in bold, higher scores reflect greater deficits)

EVERYDAY EF IN SMITH-MAGENIS AND DOWN SYNDROMES

Smith-Magenis syndrome				Down syndrome			
	<i>t</i> (df 12)	<i>p</i>	<i>d</i>		<i>t</i> (df 16)	<i>p</i>	<i>d</i>
Inhibit - Shift	4.29	.001	1.23	Inhibit > Shift	3.16	.006	.77
Inhibit – Emotional Control	2.12	.055	.60		5.39	<.001	1.31
Inhibit – Working Memory	-.51	.623	-.15		-4.83	<.001	-1.18
Inhibit – Plan/Organize	1.66	.122	.47		1.06	.305	.26
Shift - Emotional Control	-2.81	.016	-.79		2.92	.01	.72
Shift - Working Memory	-4.98	<.001	-1.46	Shift < Working Memory	-6.16	<.001	-1.50
Shift - Plan/organize	-1.33	.208	-.38		-3.21	.006	-.80
Emotional Control - Working Memory	-3.58	.004	-1.04	Emotional Control < Working Memory	-8.33	<.001	-2.02
Emotional Control -	-.18	.863	-.05		-5.46	<.001	-1.35

EVERYDAY EF IN SMITH-MAGENIS AND DOWN SYNDROMES

Plan/Organize							Plan/Organize	
Working Memory -	2.37	.039	.68		5.81	<.001	1.45	Working Memory >
Plan/Organize							Plan/Organize	